**Supplementary Table 1**: Prediction tools

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| --- | --- | --- | --- |
| **Mutation**  **Protein** | **c.1300C>T**  **p.D434Y** | **c.1525C>T**  **p.R509\*** | **c.1899\_1901del**  **p.E634del** |
| SIFT prediction  Provean score | Deleterious  -3.768 (threshold: -2.5) | Deleterious  NA | Deleterious  -9.595 (threshold: -2.5) |
| Polyphen-2 prediction  Polyphen-2 score | Probably damaging  0.993 (sensitivity: 0.47; specificity:0.96) | Probably damaging | Probably damaging |
| Mutation Taster prediction  Mutation Taster probability  Mutation Taster predicted change | Disease causing  0.99999984445111  amino acid sequence changed; protein features (might be) affected; splice site changes | Disease causing  1  NMD; amino acid sequence changed; protein features (might be) affected; splice site changes | Disease causing  0.999999975957048  amino acid sequence changed; protein features (might be) affected; splice site changes |